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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/15867
DOI URL: http://dx.doi.org/10.21474/IJAR01/15867

RESEARCH ARTICLE

BEST'S DISEASE: A CASE REPORT

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Manuscript Info

Manuscript History
Received: 10 October 2022
Final Accepted: 14 November 2022
Published: December 2022

Key words:-
Best Disease, Vitellin, Macular Dystrophy

Abstract

Best's disease is an autosomal dominant hereditary macular dystrophy characterized by the presence of auto-fluorescent vitelline deposits. We report a case of a vitelliform stage of best's disease in a 23-year-old female patient. The diagnosis was made based on the fundus appearance: egg yolk macular lesion, hypofluorescence in the early stages, OCT appearance and alteration of the electro-oculogram and the electroretinogram.

The exact frequency of this condition is difficult to determine and varies according to the various studies. It is thought to account for 4% of all retinal dystrophies. Its management is an annual surveillance.

Introduction:-

Best's disease or vitelliform macular dystrophy is an autosomal dominant hereditary macular dystrophy characterized by the presence of auto-fluorescent vitelline deposits with a stereotyped evolutionary sequence from the appearance to the fragmentation of the material until its resorption. The disease usually occurs in the first decade. A slow visual deterioration is the usual evolution of the disease, hence its incidental discovery. Choroidal neovascularization may occur in rare cases. It is also associated with panophthalmic involvement including nanophthalmos, microcornea, hyperopia and narrow anterior chamber angle with angle closure glaucoma.

Clinical observation:

We report the case of a 23-year-old female patient, the eldest of three siblings, with no particular pathological history.

The patient's best corrected visual acuity was 6/10 in the right eye and 5/10 in the left eye. Slit lamp examination revealed normam anterior segments in both eyes. The fundus examination showed a yellow macular lesion, with an aspect of "egg yolk on the plate" (Figure1). Fluorescein angiography showed early hyperfluorescence followed by hypofluorescence. Optical coherence tomography (OCT) showed an optically empty space between the neuroretina and the retinal pigment epithelium (RPE) (Figure 2).

A workup was performed: the electro-oculogram revealed an Arden coefficient of 120% in the right eye and 110%

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in the left eye.The electroretinogram showed a decrease of the amplitudes of the waves with normal times of culmination of the right eye interesting the scotopic system in favour of a Best's disease evolution.

The diagnosis of Best vitelliform macular dystrophy was made based on the appearance of the fundus, the hypofluorescence at the early stages, the OCT aspect and the alteration of the electro-oculogram and the electroretinogram. The differential diagnoses that may be evoked are Stargardt's disease, progressive cone dystrophy and X-linked retinoschisis. No treatment was proposed for this patient but simple monitoring.

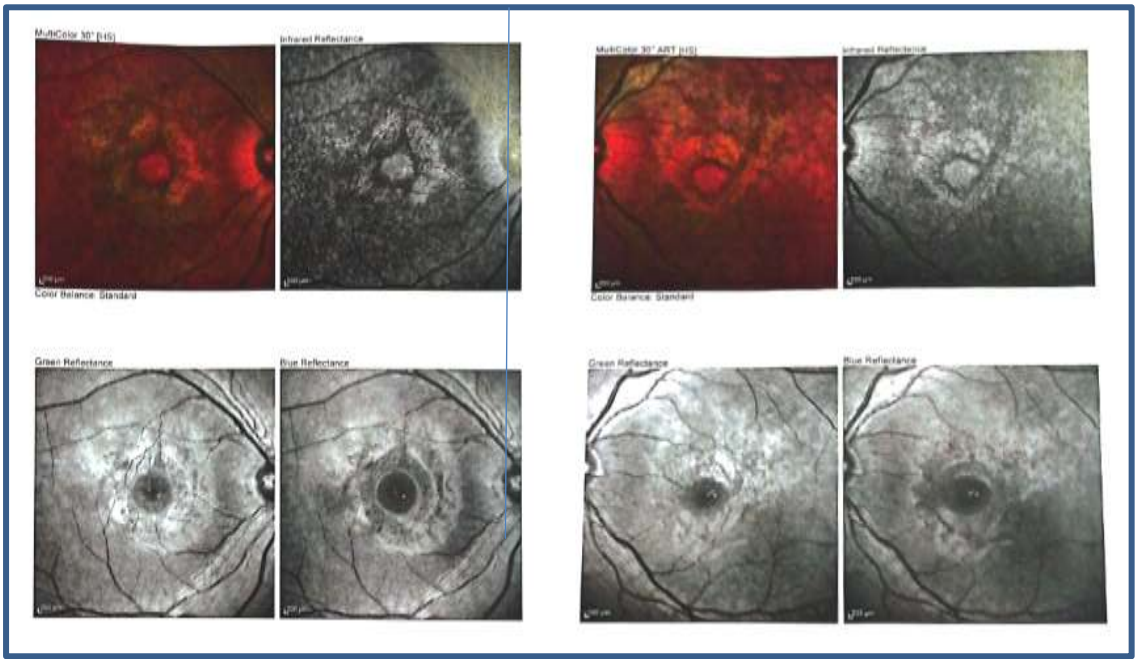

Figure 1:- Fundus images and infrared autofluorescent images of both eyes showing egg yolk appearance.

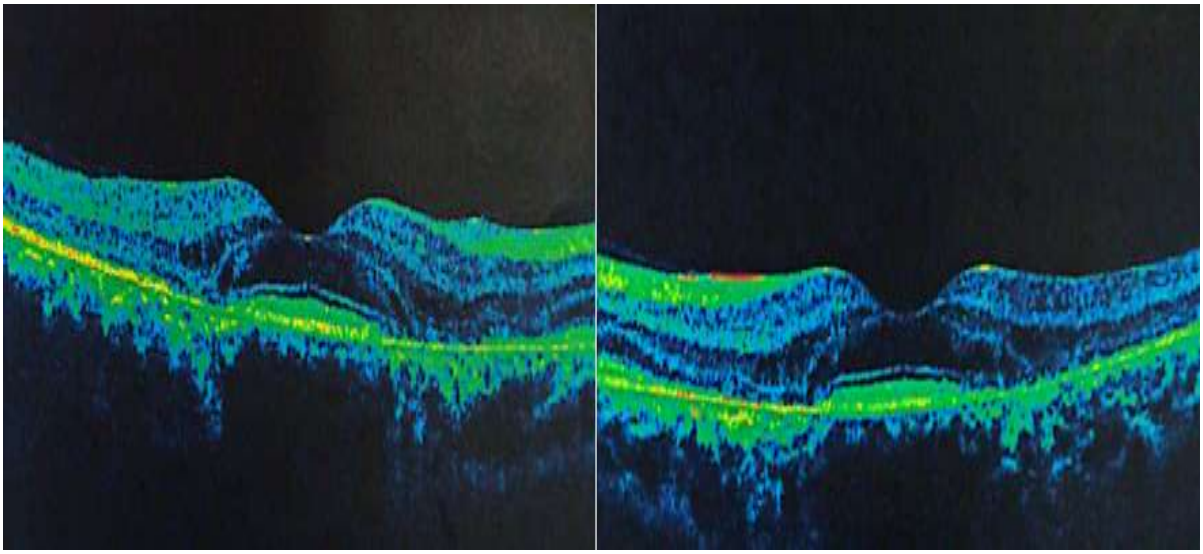

Figure 2:- Images of Macular oct of both eyes showing hyperreflective deposits in the pigment epithelium with macular elevation.

Discussion:-

In 1905, Best described all stages of hereditary macular dystrophy from eight related patients in a large family of 59 individuals and gave the disease its name [1]. Zanen likened the lesion to an "egg-on-the-fly" appearance and

introduced the term vitelliform disc in 1950 [2]. The exact frequency of the condition is difficult to determine and varies according to the various studies. It is thought to account for 4% of all retinal dystrophies [3]. Best's disease is transmitted exclusively in the autosomal dominant mode. Penetrance is incomplete, expressivity is variable and many cases are sporadic. In 1992, the gene for Best's disease was located on the long arm of chromosome 11, at 11q12-q13 [4]. This gene was then identified: the VMD2 (vitelliform macular dystrophy-2) gene, known as bestrophin [5]. There are several mutations of this gene that are responsible for Best's disease [6]. The bestrophin protein plays a role in transmembrane ion channels [7].
The age of onset is most often between 7 and 12 years. The most frequent circumstances of discovery are a decrease in central visual acuity, uni- or bilateral, visual blur, and metamorphopsia. However, the discovery of a vitelliform lesion on fundus examination may be incidental or part of a family investigation [8,9].Before the appearance of the typical vitelliform lesion, it is difficult to individualize a macular lesion.

There are several clinical and angiographic stages: pre-vitelliform stage, vitelliform stage, remodeling stage, atrophic stage and fibroglial stage. The electro-oculogram (EOG) is a fundamental examination for the diagnosis of Best's disease [10, 11], with an Arden ratio lower than 145. In addition, EOG can be used at a preclinical stage and may preclinical stage and may be contributory. Optical coherence tomography (OCT), a relatively recent development, is contributory both for diagnosis and for patient follow-up with different aspects depending on the stage of evolution [12]. In the vitelliform stage, there is a raised appearance just below the plane of the pigment epithelium, with hyperreflectivity of the pigment epithelium layer. The innermost layers are spared. The electroretinogram (ERG) is normal in Best's disease in the vitelliform stage. At the later stage of the evolution, atrophic or fibroglial stage, the foveolar ERG can be disturbed.

Conclusion:-

The semiological analysis of the fundus remains the fundamental element of the diagnosis. The fundus examination, associated with the clinical history and the family history, will lead to the request of additional targeted retinal or electroretinographic imaging. Management is simple surveillance with referral to low vision centers when visual acuity deteriorates.

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